SELENON gene

selenoprotein N

Normal Function

The *SELENON* gene (also called *SEPN1*) provides instructions for making a protein called selenoprotein N. This protein is part of a family of selenoproteins, which have several critical functions within the body. Selenoproteins are primarily involved in chemical reactions called oxidation-reduction reactions, which are essential for protecting cells from damage caused by unstable oxygen-containing molecules. Selenoprotein N is likely involved in protecting cells against oxidative stress. Oxidative stress occurs when unstable molecules called free radicals accumulate to levels that damage or kill cells.

The exact function of selenoprotein N is unknown. This protein is highly active in many tissues before birth and may be involved in the formation of muscle tissue (myogenesis). Selenoprotein N may also be important for normal muscle function after birth, although it is active at much lower levels in adult tissues. This protein contains a region that likely allows it to bind to calcium. This region is of interest because calcium plays an important role in triggering muscle contractions, which allow the body to move.

Health Conditions Related to Genetic Changes

multiminicore disease

At least 17 mutations in the *SELENON* gene have been identified in people with the classic form of multiminicore disease. Many of these genetic changes lead to the production of an abnormally short version of selenoprotein N. Other mutations change single protein building blocks (amino acids) in critical regions of the protein. The effects of changes in the structure and function of selenoprotein N are unknown, and researchers are working to determine how these changes lead to muscle weakness and the other characteristic features of multiminicore disease.

other disorders

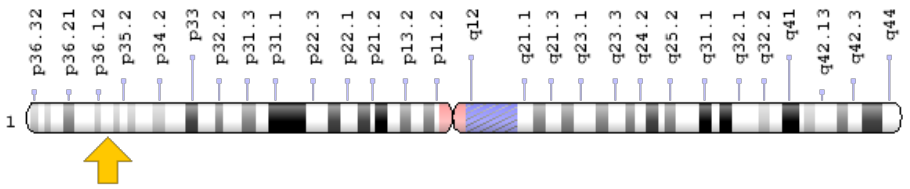
Mutations in the *SELENON* gene are responsible for several other rare muscle disorders, including rigid spine muscular dystrophy and desmin-related myopathy with Mallory body-like inclusions. These conditions cause muscle weakness, particularly in the muscles of the trunk and neck. Affected individuals also have decreased muscle tone (hypotonia), abnormal curvature of the spine (scoliosis), and serious breathing problems. Because they have a similar pattern of signs and symptoms and are caused by mutations in the same gene, many researchers

believe that these conditions are all part of a single syndrome with variable signs and symptoms. Together, muscle diseases caused by *SELENON* gene mutations are known as *SELENON*-related (or *SEPN1*-related) myopathy.

Chromosomal Location

Cytogenetic Location: 1p36.11, which is the short (p) arm of chromosome 1 at position 36.11

Molecular Location: base pairs 25,800,176 to 25,818,222 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- selenoprotein N, 1
- SELN
- SEPN1
- SEPN1_HUMAN

Additional Information & Resources

Educational Resources

- Muscular Dystrophy UK
  http://www.musculardystrophyuk.org/

GeneReviews

- Congenital Fiber-Type Disproportion
  https://www.ncbi.nlm.nih.gov/books/NBK1259
- Congenital Muscular Dystrophy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1291
- Multiminicore Disease
  https://www.ncbi.nlm.nih.gov/books/NBK1290

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SEPN1%5BTIAB%5D%29+OR+%28selenoprotein+N,+1%5BTIAB%5D%29%29+OR+%28SELN%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- SELENOPROTEIN N
  http://omim.org/entry/606210

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SELENON.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SELENON%5Bgene%5D
- HGNC Gene Family: EF-hand domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/863
- HGNC Gene Family: Selenoproteins
  http://www.genenames.org/cgi-bin/genefamilies/set/890
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15999
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/57190
- UniProt
  http://www.uniprot.org/uniprot/Q9NZV5

Sources for This Summary

- Arbogast S, Beuvin M, Fraysse B, Zhou H, Muntoni F, Ferreiro A. Oxidative stress in SEPN1-related myopathy: from pathophysiology to treatment. Ann Neurol. 2009 Jun;65(6):677-86. doi: 10.1002/ana.21644.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19557870
- Clarke NF, Kidson W, Quijano-Roy S, Estournet B, Ferreiro A, Guicheney P, Manson JI, Kornberg AJ, Shield LK, North KN. SEPN1: associated with congenital fiber-type disproportion and insulin resistance. Ann Neurol. 2006 Mar;59(3):546-52.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16365872

- Ferreiro A, Ceuterick-de Groote C, Marks JJ, Goemans N, Schreiber G, Hanefeld F, Fardeau M, Martin JJ, Goebel HH, Richard P, Guicheney P, Bönnemann CG. Desmin-related myopathy with Mallory body-like inclusions is caused by mutations of the selenoprotein N gene. Ann Neurol. 2004 May;55(5):676-86.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15122708
- Ferreiro A, Quijano-Roy S, Pichereau C, Moghadaszadeh B, Goemans N, Bönnemann C, Jungbluth H, Straub V, Villanova M, Leroy JP, Romero NB, Martin JJ, Muntoni F, Voit T, Estournet B, Richard P, Fardeau M, Guicheney P. Mutations of the selenoprotein N gene, which is implicated in rigid spine muscular dystrophy, cause the classical phenotype of multiminicore disease: reassessing the nosology of early-onset myopathies. Am J Hum Genet. 2002 Oct;71(4):739-49. Epub 2002 Aug 21.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12192640
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC378532/
- GeneReview: Congenital Fiber-Type Disproportion
  https://www.ncbi.nlm.nih.gov/books/NBK1259
- GeneReview: Multiminicore Disease
  https://www.ncbi.nlm.nih.gov/books/NBK1290
- Jungbluth H. Multi-minicore Disease. Orphanet J Rare Dis. 2007 Jul 13;2:31. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17631035
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1947955/
- Lescure A, Rederstorff M, Krol A, Guicheney P, Allamand V. Selenoprotein function and muscle disease. Biochim Biophys Acta. 2009 Nov;1790(11):1569-74. doi: 10.1016/j.bbagen.2009.03.002. Epub 2009 Mar 11. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19285112
- Okamoto Y, Takashima H, Higuchi I, Matsuyama W, Suehara M, Nishihira Y, Hashiguchi A, Hirano R, Ng AR, Nakagawa M, Izumo S, Osame M, Arimura K. Molecular mechanism of rigid spine with muscular dystrophy type 1 caused by novel mutations of selenoprotein N gene. Neurogenetics. 2006 Jul;7(3):175-83. Epub 2006 Jun 15.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16779558
- Petit N, Lescure A, Rederstorff M, Krol A, Moghadaszadeh B, Wewer UM, Guicheney P. Selenoprotein N: an endoplasmic reticulum glycoprotein with an early developmental expression pattern. Hum Mol Genet. 2003 May 1;12(9):1045-53.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12700173
- Schara U, Kress W, Bönnemann CG, Breitbach-Faller N, Korenke CG, Schreiber G, Stoetter M, Ferreiro A, von der Hagen M. The phenotype and long-term follow-up in 11 patients with juvenile selenoprotein N1-related myopathy. Eur J Paediatr Neurol. 2008 May;12(3):224-30. Epub 2007 Oct 22.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17951086
- Zorzato F, Jungbluth H, Zhou H, Muntoni F, Treves S. Functional effects of mutations identified in patients with multiminicore disease. IUBMB Life. 2007 Jan;59(1):14-20. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17365175

Reviewed: July 2010
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services